

## Agenda

Friday, March 3, 2017

8:00-8:30	<b>Registration and Continental Breakfast Provided</b>	11:45-12:45	<b>KEYNOTE ADDRESS</b> <b>Genotype Does Not Always Explain Phenotype for Skeletal Disorders</b> Debbie Krakow, MD David Geffen School of Medicine, UCLA
8:30-8:35	<b>Welcome</b> Bruce R. Korf, MD, PhD		
<b>SESSION 1</b> 8:35-10:00	<b>INDICATIONS FOR REFERRAL</b> Nathaniel Robin, MD - Moderator	12:45-1:00	<b>Break</b>
8:35-9:10	<b>Big Size, Small Size: Genetic Causes of Growth Differences</b> Nathaniel Robin, MD University of Alabama at Birmingham	<b>LUNCH/ SESSION 3</b> 1:00-2:00	<b>PARENT PANEL</b> Ashley Cannon, PhD, MS, CGC - Moderator University of Alabama at Birmingham
9:10-9:45	<b>Physical Clues to Explain Developmental Differences</b> Chad Haldeman-Englert, MD Mission Health, Asheville, NC	<b>SESSION 4</b> 2:00-3:30	<b>OUTCOMES AND FOLLOW-UP</b> Bruce R. Korf, MD, PhD - Moderator
9:45-10:00	<b>Questions and Panel Discussion</b> NHR, CH-E, ACHE	2:00-2:30	<b>Update on the UAB Undiagnosed Diseases Program</b> Bruce R. Korf, MD, PhD University of Alabama at Birmingham
10:00-10:15	<b>Break</b>	2:30-3:00	<b>Outcomes of Genetic Counseling Following Genome Sequencing: Experience From the HudsonAlpha Clinical Sequencing Exploratory Research project</b> Whitley Kelley, MS HudsonAlpha, Huntsville, AL
<b>SESSION 2</b> 10:15-11:45	<b>DIAGNOSTIC APPROACHES</b> Anna C.E. Hurst, MD, MS - Moderator		
10:15-10:45	<b>Cytogenetic Testing in the Genetic Evaluation</b> Fady Mikhail, MD, PhD University of Alabama at Birmingham	3:00-3:30	<b>Ethical and Legal Issues in Diagnostic Genome Sequencing</b> Tom May, PhD HudsonAlpha, Huntsville, AL
10:45-11:25	<b>Molecular Genomic Testing Strategies</b> David Bick, MD HudsonAlpha, Huntsville, AL		
11:25-11:45	<b>Practical Resources for the Pediatrician</b> Anna C.E. Hurst, MD, MS University of Alabama at Birmingham		



## About Our Keynote Speaker

Debbie Krakow, MD  
Professor, Orthopaedic Surgery, Human Genetics  
and Obstetrics and Gynecology  
David Geffen School of Medicine, UCLA  
Los Angeles, CA

Dr. Krakow is an internationally recognized expert on the diagnosis and management of skeletal disorders. She is Professor of Human Genetics and Orthopaedic Surgery at UCLA. She also is Co-Director of the UCLA International Skeletal Dysplasia Registry, a referral center for that focuses on clinical diagnoses and the molecular biology of genetic bone diseases. In addition to her clinical work as a medical geneticist and maternal fetal medicine physician, Dr. Krakow directs a laboratory that has identified the genetic and cellular basis of skeletal diseases. She has authored over 100 peer reviewed papers, numerous reviews and book chapters.

## Learning Objectives

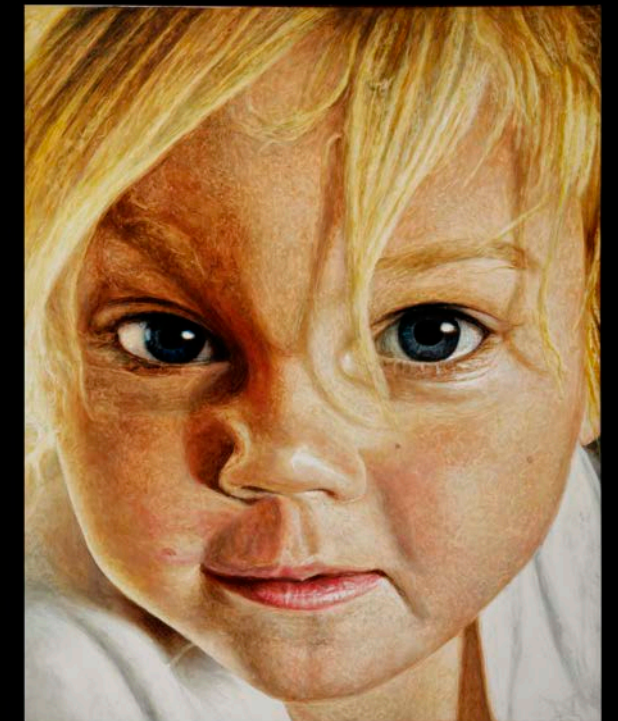
UPON COMPLETION OF THIS LIVE ACTIVITY,  
PARTICIPANTS WILL BE ABLE TO:

- Recognize features of a child who may benefit from a genetic evaluation.
- Identify the benefits and limitations of chromosomal microarray array testing.
- Distinguish when a child may benefit from broad molecular-based testing strategies (such as whole exome or whole genome sequencing).
- Utilize practical resources for the healthcare management of a child with a genetic condition.
- Reflect on the psychosocial needs of the family of a child with a genetic condition.
- Review outcomes from existing genetic programs in the region.

## Continuing Education

Children's of Alabama designates this live activity for a maximum of 5.25 AMA PRA Category 1 Credits™. Physicians should claim only the credit commensurate with the extent of their participation in the activity.

Children's of Alabama is accredited by the Medical Association of the State of Alabama to provide continuing medical education for physicians.



### BEYOND THE DIAGNOSIS ART EXHIBIT

Selected works from this traveling art exhibit, focusing on the rare disease patient, will be displayed at Children's of Alabama from February 20 through March 17, 2017. Art has been used for thousands of years to successfully convey a message, whether it be a story or a glimpse into the human spirit. The purpose of this exhibit, presented by the Rare Disease United Foundation, is to encourage a look "beyond the diagnosis" to the patient. Two works from the exhibit are shown in this brochure, including the one above — "Ashlyn" (STURGE-WEBER SYNDROME) by Wolfgang Widmoser.

## Registration

FOR ONLINE REGISTRATION, please visit  
**ChildrensAL.org/genetics**

Or complete the form below and mail, email or fax  
registration form and payment to:  
Shaila Handattu, PhD, MBA  
Department of Genetics  
Kaul 230, 720 20th Street South  
Birmingham, AL 35294  
hande@uab.edu FAX 205.934.9488

**\$35 to Attend**

Please make checks payable to Children's of Alabama.

NAME \_\_\_\_\_

ADDRESS \_\_\_\_\_

PHONE \_\_\_\_\_

FAX \_\_\_\_\_

EMAIL \_\_\_\_\_

\_\_\_\_ PHYSICIAN      \_\_\_\_ NURSE

\_\_\_\_ Check here if a Children's or UAB employee.  
Symposium is free to Children's of Alabama  
and UAB employees, but you must show your  
ID badge to enter.

Special dietary needs \_\_\_\_\_

**Parking will be available  
in the Children's Park Place Deck.**

The entrance to the deck is at  
16th Street South and 5th Avenue South.

**Questions?**

Contact Shaila Handattu at hande@uab.edu

## Speakers

**David Bick, MD**

Medical Director, HudsonAlpha Medical Genomics Program  
Huntsville, AL

**Ashley Cannon, PhD, MS, CGC**

Instructor, Certified Genetic Counselor  
Department of Genetics  
University of Alabama at Birmingham (UAB)

**Chad Haldeman-Englert, MD**

Clinical Geneticist, Fullerton Genetics Center  
Mission Health, Asheville, NC

**Anna Chesson Edens Hurst, MD, MS**

Assistant Professor, Department of Genetics  
University of Alabama at Birmingham (UAB)

**Whitley V. Kelley, MS**

Genetic Counselor, Clinical Applications Specialist  
HudsonAlpha Institute for Biotechnology  
Huntsville, AL

**Bruce R. Korf, MD, PhD**

Professor and Chair, Department of Genetics  
Director, Heflin Center for Genomic Sciences  
Co-Director, UAB-HudsonAlpha Center for Genomic Medicine  
University of Alabama at Birmingham (UAB)

**Deborah Krakow, MD**

Professor, Orthopaedic Surgery, Human Genetics and  
Obstetrics and Gynecology  
David Geffen School of Medicine, UCLA  
Los Angeles, CA

**Thomas May, PhD**

Senior Scientist  
HudsonAlpha Institute for Biotechnology  
Huntsville, AL

**Fady M. Mikhail, MD, PhD**

Associate Professor and Co-Director  
Clinical Cytogenetics Laboratory, Department of Genetics  
University of Alabama at Birmingham (UAB)

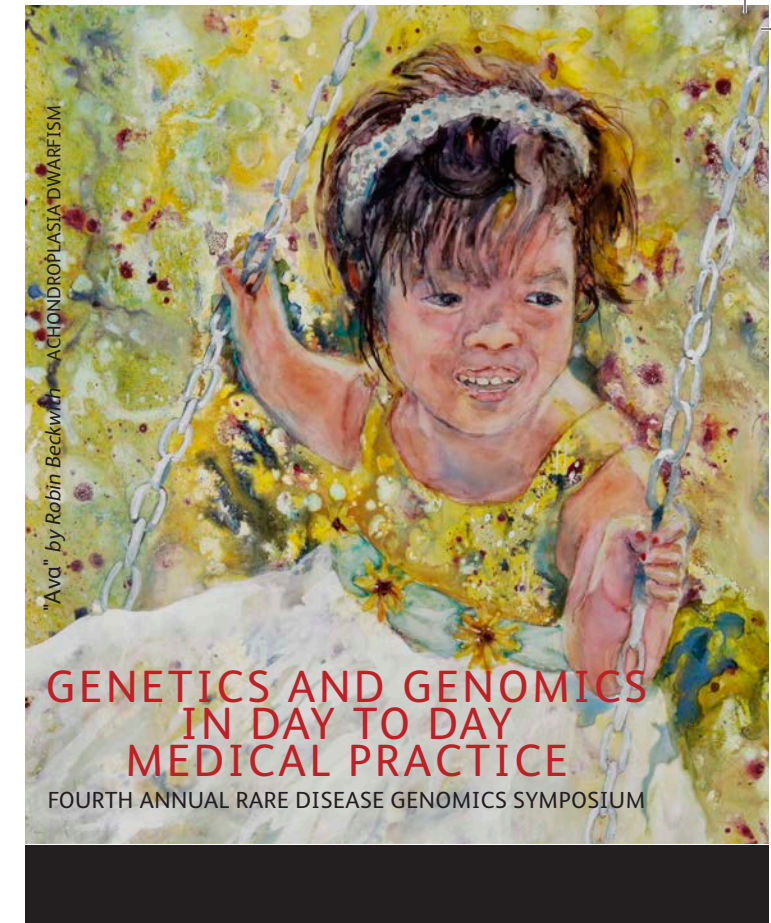
**Nathaniel H. Robin, MD**

Professor of Genetics and Pediatrics  
University of Alabama at Birmingham (UAB)



Children's  
of Alabama®

Corporate Communications & Marketing  
1600 7th Avenue South  
Birmingham, AL 35233



March 3, 2017

Friday, 8:00am — 3:30pm

Bradley Lecture Center

4th Floor, Children's Harbor Building

1600 6th Avenue South

Birmingham, AL 35233



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